

What is Claimed:

1. A purified and isolated wild type *PKD2* nucleic acid.
2. The nucleic acid of Claim 1 which is genomic DNA, cDNA or RNA.
3. The nucleic acid of Claim 1 encoding the amino acid sequence contained in Figure 5.
4. The nucleic acid of Claim 1 having the nucleotide sequence contained in Figure 5.
5. A purified and isolated mutated *PKD2* nucleic acid.
6. The mutated *PKD2* nucleic acid of Claim 5 which has one or more deletion, insertion, point or rearrangement mutations.
7. A single-stranded nucleic acid probe which specifically hybridizes to wild type *PKD2* nucleic acid.
8. The probe of Claim 7 which is labeled with a detectable marker.
9. A mixture of single-stranded nucleic acid probes each of which specifically hybridizes to wild type *PKD2* nucleic acid.
10. A single-stranded nucleic acid probe which specifically hybridizes to mutated *PKD2* nucleic acid.
11. The probe of Claim 10 which is labeled with a detectable marker.
12. A mixture of single-stranded nucleic acid probes each of which specifically hybridizes to mutated *PKD2* nucleic acid.
13. A kit useful for detecting wild type *PKD2* nucleic acid comprising one or more single-stranded nucleic acid probes which specifically hybridize to wild type *PKD2* nucleic acid.
14. A kit useful for detecting mutated *PKD2* nucleic acid comprising one or more single-stranded nucleic acid probes which specifically hybridize to mutated *PKD2* nucleic acid.

15. A kit useful for detecting wild type *PKD2* nucleic acid and mutated *PKD2* nucleic acid comprising (a) one or more single-stranded nucleic acid probes which specifically hybridize to wild type *PKD2* nucleic acid, and (b) one or more single-stranded nucleic acid probes which specifically hybridize to mutated *PKD2* nucleic acid.

16. A vector comprising a nucleic acid encoding an active *PKD2* protein.

17. The vector of Claim 16 wherein the *PKD2* protein is the wild type protein or an analogue thereof.

18. The vector of Claim 16 wherein the nucleic acid encodes the amino acid sequence contained in Figure 5.

19. The vector of Claim 16 wherein the nucleic acid has the nucleotide sequence contained in Figure 5.

20. A cell stably transformed with a vector comprising a nucleic acid encoding an active *PKD2* protein.

21. The cell of Claim 20 wherein the *PKD2* protein is the wild type protein or an analogue thereof.

22. The cell of Claim 21 wherein the nucleic acid encodes the amino acid sequence contained in Figure 5.

23. The cell of Claim 21 wherein the nucleic acid has the nucleotide sequence contained in Figure 5.

24. The cell of Claim 20 which is eukaryotic.

25. The cell of Claim 20 which is prokaryotic.

26. A method for producing a recombinant, active *PKD2* protein comprising culturing a cell transformed with a vector comprising a nucleic acid encoding an active *PKD2* protein, and recovering *PKD2* protein from the culture.

27. The method of Claim 26 wherein the *PKD2* protein is the wild type protein or an analogue thereof.

28. The method of Claim 26 wherein the nucleic acid encodes the amino acid sequence contained in Figure 5.

29. The method of Claim 26 wherein the nucleic acid has the nucleotide sequence contained in Figure 5.

30. The method of Claim 26 wherein the cell is eukaryotic.

31. The method of Claim 26 wherein the cell is prokaryotic.

32. A purified, active PKD2 protein.

33. The protein of Claim 32 which is the wild type protein or an analogue thereof.

34. The protein of Claim 32, which is recombinantly produced.

35. The protein of Claim 32 which has the amino acid sequence contained in Figure 5.

36. An antibody immunoreactive with a wild type PKD2 protein or an analogue thereof.

37. The antibody of Claim 36 which is polyclonal.

38. The antibody of Claim 36 which is monoclonal.

39. The antibody of Claim 36 which is labeled with a detectable marker.

40. An antibody immunoreactive with a mutant PKD2 protein.

41. The antibody of Claim 40 which is polyclonal.

42. The antibody of Claim 40 which is monoclonal.

43. The antibody of Claim 40 which is labeled with a detectable marker.

44. A kit useful for detecting wild type PKD2 protein comprising an antibody immunoreactive with wild type PKD2 protein.

45. A kit useful for detecting mutant PKD2 protein comprising an antibody immunoreactive with mutant PKD2 protein.

46. A kit useful for detecting wild type PKD2 protein and mutant PKD2 protein comprising an antibody immunoreactive with wild type PKD2 protein and an antibody immunoreactive with mutant PKD2 protein.

47. A method for diagnosing autosomal dominant polycystic kidney disease caused by a mutated *PKD2* gene in a subject comprising detecting the presence of a mutated *PKD2* gene in nucleic acid of the subject.

48. The method of Claim 47 wherein the subject is an embryo, fetus, newborn, infant or adult.

49. The method of Claim 47 wherein the mutated *PKD2* gene has one or more deletion, insertion, point or rearrangement mutations.

50. The method of Claim 47 wherein the nucleic acid is DNA or RNA.

51. The method of Claim 47 wherein the presence of the mutated *PKD2* gene is detected by one or more techniques selected from the group consisting of sequence analysis, restriction enzyme digestion analysis, hybridization and polymerase chain reaction.

52. The method of Claim 47 wherein the presence of the mutated *PKD2* gene is detected by the presence of a gene product encoded by the mutated *PKD2* gene.

53. The method of Claim 52 wherein the gene product is a protein.

54. The method of Claim 53 wherein the protein is detected by immunoblotting, immunoprecipitation, solid phase radioimmunoassay, or enzyme-linked immunoabsorbent assay.

55. The method of Claim 52 wherein the gene product is mRNA.

56. The method of Claim 55 wherein the mRNA is detected by one or more techniques selected from the group consisting of sequence analysis, restriction enzyme digestion analysis, hybridization and polymerase chain reaction.

57. A method for treating autosomal dominant polycystic kidney disease caused by a mutated *PKD2* gene in a subject in need of such treatment comprising the delivery and expression of a functional *PKD2* gene into a sufficient number of cells of the subject to treat the disease.

58. The method of Claim 57 wherein the subject is an embryo, fetus, newborn, infant or adult.

59. The method of Claim 57 wherein the functional PKD2 gene is delivered by homologous recombination.

60. The method of Claim 57 wherein the functional PKD2 gene is delivered by a vector.

61. The method of Claim 60 wherein the vector is a viral vector.

62. The method of Claim 61 wherein the viral vector is a retrovirus.

63. The method of Claim 61 wherein the viral vector is a DNA virus.

64. The method of Claim 57 wherein the functional PKD2 gene is delivered by transfection.

65. A recombinant viral vector for treating a defect in a *PKD2* gene in a target cell comprising (a) the nucleic acid of or corresponding to at least a portion of the genome of a virus, which portion is capable of directing the infection of the target cell, and (b) a *PKD2* gene operably linked to the viral nucleic acid and capable of being expressed as a functional gene product in the target cell.

66. The recombinant viral vector of Claim 65 which is a retrovirus.

67. The recombinant viral vector of Claim 65 which is a DNA virus.

68. A stem cell which expresses a functional PKD2 gene introduced therein through viral transduction.

69. A stem cell which expresses a functional PKD2 gene introduced therein through homologous recombination.

70. A stem cell which expresses the functional PKD2 gene introduced therein through transfection.

71. A vector comprising a mutated *PKD2* gene which is capable of introducing the mutated *PKD2* gene in at least some embryonic cells to which the vector is introduced.

72. An embryonic stem cell comprising a mutated *PKD2* gene which has been integrated into the cell following transduction with the vector of Claim 71.

73. A non-human, transgenic animal comprising a mutated *PKD2* gene.

74. The non-human, transgenic animal of Claim 73 whose germ and somatic cells contain a mutated *PKD2* gene sequence introduced into said animal, or an ancestor thereof, at an embryonic stage.

75. A method of producing a non-human, transgenic animal comprising introducing a mutated *PKD2* gene into at least some cells of a recipient, non-human animal.